First Screen, Not Worst Screen! (The Role of the First Trimester Screen in the face of Normal Cell Free DNA)

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Background

- Cell free DNA (cfDNA) has emerged in popularity due to its high detection rates for Trisomy 13, 18, and 21.
- However, there is no consensus as to which screening method for aneuploidy should be offered.
- American College of Obstetricians and Gynecologists recommends a first trimester screen (FTS) or cfDNA should be offered to all patients, and do not recommend multiple screening tests. Per the Society for Maternal-Fetal Medicine, measuring the nuchal translucency (NT) in women with normal cfDNA provides limited additional benefit. The American College of Medical Genetics recommends patient centered counseling regarding screening and diagnostic approaches available.

Objective

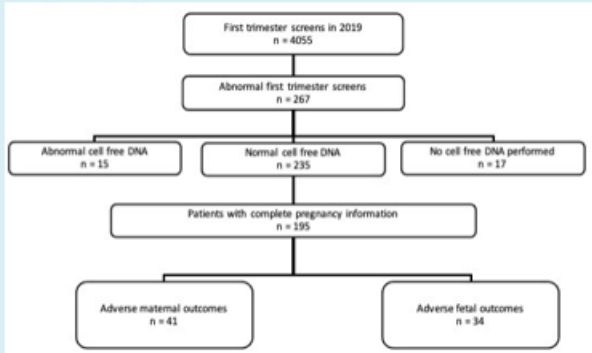
- It is common practice in our hospital to perform FTS, inclusive of NT and serum analytes, with or without cfDNA.
- The aim of this study was to assess the clinical relevance of an **abnormal** FTS in a cohort of women with **normal** cell free DNA, with respect to maternal and fetal pregnancy outcomes.

Study Design

- Retrospective descriptive chart review was performed for pregnant women who underwent FTS at MSW from 1/2019-12/2019
- Included patients had abnormal FTS; consisting of NT >3 mm or >95 percentile for gestational age, PAPP-A <0.4 MoM, bHCG >2.5 MoM, or overall increased risk for Trisomy 13, 18, or 21.
- Patients with abnormal cfDNA, cfDNA not performed, or incomplete pregnancy data were excluded.

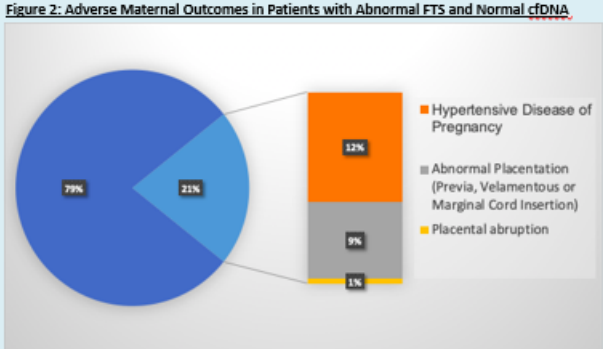
Results

Figure 1: Study Design



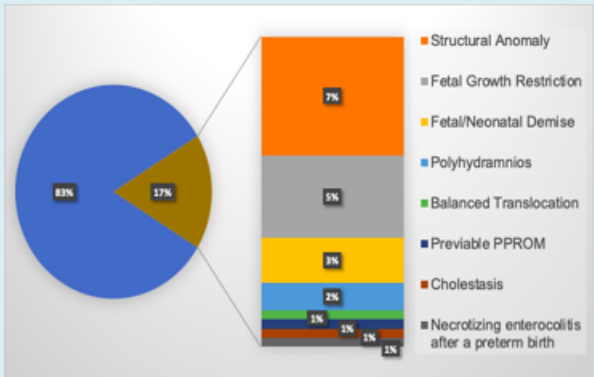
- Of the 195 patients with complete records and normal cfDNA, 35 (17%) had low PAPP-A, 34 (17%) had increased NT, 10 (5%) had a high bHCG, and 172 (88%) had an overall increased risk on their FTS.

Figure 2: Adverse Maternal Outcomes in Patients with Abnormal FTS and Normal cfDNA



Results

Figure 3: Adverse Maternal Outcomes in Patients with Abnormal FTS and Normal cfDNA



- 21% of women had adverse maternal outcomes including hypertension, abnormal placentation (previa, velamentous and/or marginal cord insertion), and placental abruption.
- 17% of women had adverse fetal outcomes including fetal growth restriction, structural anomalies, fetal demise, polyhydramnios, previable PPROM, necrotizing enterocolitis, and a balanced translocation.

Conclusions

- Our data suggests that the practice of performing universal FTS with cell free DNA may have a clinical benefit.
- Screening with only cfDNA may limit opportunities for early diagnosis and/or intervention for other maternal and fetal outcomes
- We suggest that patients with an abnormal FTS may benefit from increased vigilance in prenatal care.